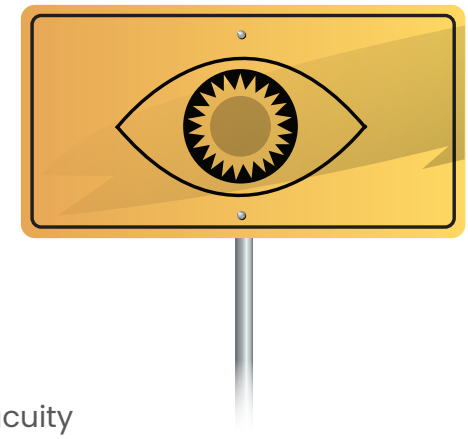
Case Studies: Cerebrotendinous Xanthomatosis (CTX)

For US HCPs only

Case study 1: A 9-year-old child with bilateral cataracts[1]

Patient referred to an ophthalmologist



Presenting symptoms

- Decreased vision over several months

Medical history

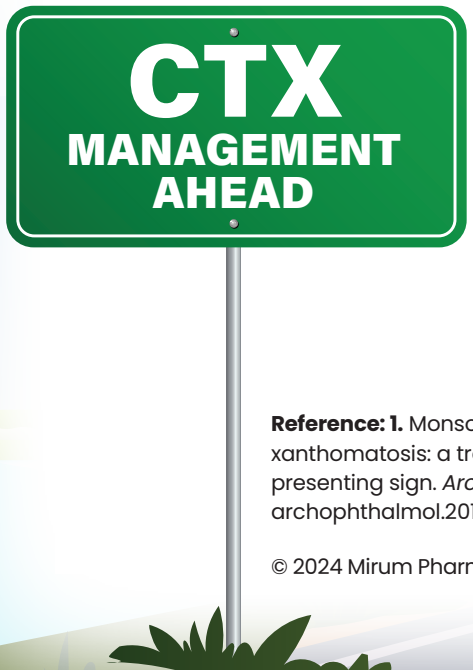
- Significant for diarrhea and nonverbal learning disorder

Examination

- Uncorrected distance visual acuity: 20/80 OU
- Uncorrected near acuity: 20/50 OU
- Manifest refraction: 0.75+0.50 X 90 OU, with a corrected distance acuity of 20/60 OD and 20/80 OS
- Complete dilated examination: remarkable for the presence of a cataract in each eye, described as a diffuse nuclear haze on slit lamp examination, with mild posterior capsular opacification
- Metabolic workup: plasma 5-cholestanol level was markedly elevated at 3.7 mg/dL (normal <0.2 mg/dL), which was **suggestive of CTX**

Outcome

- Sequential bilateral cataract surgery with intraocular lens placement corrected visual acuity to 20/20 OU
- After 6 years, due to early diagnosis and appropriate management, no further clinical manifestations of CTX had developed



Reference: 1. Monson DM, DeBarber AE, Bock CJ, et al. Cerebrotendinous xanthomatosis: a treatable disease with juvenile cataracts as a presenting sign. *Arch Ophthalmol.* 2011;129(8):1087-1088. doi:10.1001/archophthalmol.2011.219

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Case study 2: A 55-year-old with tendon xanthomas[2]

Presenting symptoms

- Admitted to the hospital for investigations relating to a slowly progressive paraparesia and joint deformities

Medical and family history

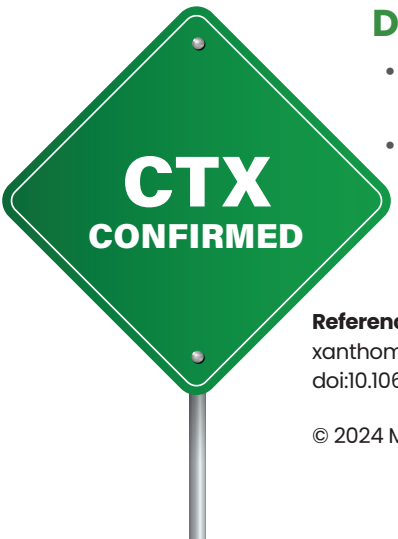
- Difficulty standing and walking since infancy
- Bilateral juvenile cataracts
- Unable to complete education because of intellectual disability
- 3 siblings died in childhood, one from an undiagnosed neurological disorder
- Unknown whether patient's parents were related

Physical and neurological examination

- Physical examination:
  - Tendon xanthomas—firm, round, noninflammatory, subcutaneous tumors measuring 6 cm in diameter over the knees and elbows, which adhered to tendons
  - Yellowish papules of 2 to 3 mm in diameter in the superior eyelid, compatible with xanthelasmas
- Neurological findings included:
  - Mental retardation, spastic-ataxic gait, bilateral Babinski sign, symmetric amyotrophia on inferior and superior extremities, and hyperactive deep tendon reflexes with associated left Achilles clonus
- Ligamentous hyperlaxity was observed
- Electroencephalogram showed a mild disorganization of the basic activity of theta waves

Laboratory testing and imaging studies

- Standard laboratory test values were normal
- Biopsy from the left knee:
  - Infiltrate of foam cells surrounded by fibrous tracts
  - Inflammatory cells such as lymphocytes, histiocytes, and neutrophils were observed around the foam cells in some areas, and a cholesterol cleft was found
- Magnetic resonance imaging:
  - Cerebral and cerebellar atrophy and hyperintense signals in the mesencephalic peduncles, protuberance, and cerebellar hemispheres



Diagnosis/outcome

- Because of these signs and symptoms, CTX was suspected and later confirmed by laboratory testing
- Appropriate management was initiated; however, due to the advanced state of the patient's disease, only slight improvement in spasticity was noted

Reference: 2. Bel S, García-Patos V, Rodríguez L, et al. Cerebrotendinous xanthomatosis. *J Am Acad Dermatol*. 2001;45(2):292-295. doi:10.1067/mjd.2001.113690